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(54) Title: LAFORA'S DISEASE GENE

(57) Abstract

A novel gene (EPM2A) that is deleted or mutated in people with Lafora's disease is described. The EPM2A gene encodes a protein having an active catalytic site of a protein tyrosine phosphatase. Many different sequence mutations as well as several microdeletions in EPM2A have been found that co-segregate with Lafora's disease.

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# INTERNATIONAL SEARCH REPORT

Interi. ..nal Application No
PCT/CA 99/00646

A. CLASSIFICATION OF SUBJECT MATTER
IPC 7 C12Q1/68 C12N9/16 C12N15/55 C07K14/47

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)
IPC 7 C12Q

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

C. DOCUMENTS CONSIDERED TO BE RELEVANT

| Category ° | Citation of document, with indication, where appropriate, of the relevant passages | Relevant to claim No. |
|---|---|---|
| X | EMBL Database Entry HS466P17<br>ccession number AL023806; 8 June 1998<br>Thorpe K.:"Human DNA sequence from clone 466P17 on chromosome 6q24: Contains a putative novel gene, the 5' part of the EPM2A (Laforin) gene...and a ca repeat polymorphism".<br>XP002127924<br>see database entry<br>--- | 1-9, 17-19 |
| P,X | MINASSIAN B ET AL: "Mutations in a gene encoding a novel protein tyrosine phosphatase cause progressive myoclonus epilepsy"<br>NATURE GENETICS,<br>vol. 20, no. 2, October 1998 (1998-10), pages 171-74, XP000869599<br>the whole document<br>---<br>-/-- | 1-19 |

[X] Further documents are listed in the continuation of box C.

[ ] Patent family members are listed in annex.

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| European Patent Office, P.B. 5818 Patentlaan 2<br>NL - 2280 HV Rijswijk<br>Tel. (+31-70) 340-2040, Tx. 31 651 epo nl,<br>Fax: (+31-70) 340-3016 | Osborne, H |

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Inter ..onal Application No
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C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

| Category | Citation of document, with indication,where appropriate, of the relevant passages | Relevant to claim No. |
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| P,X | SERRATOSA J ET AL: "A novel protein tyrosine phosphatase gene is mutated in progressive myoclonus epilepsy of the Lafora type (EPM2)" HUMAN MOLECULAR GENETICS, vol. 8,. no. 2, February 1999 (1999-02), pages 345-52, XP000867126 the whole document | 1-19 |
| A | MINASSIAN B ET AL : "Progesss towards the positional cloning of a gene for Lafora's disease" NEUROLOGY , vol. 48, no. 3 suppl 2, 12 - 19 April 1997, page A428 XP000866505 see abstract P06.091 | 1-19 |
| A | SAINZ J ET AL: "Lofora's progressive myoclonus epilepsy : narrowing the chromosome 6Q24 locus by recombinations and homozygosities" AMERICAN JOURNAL OF HUMAN GENETICS, vol. 61, no. 5, 1997, pages 1205-09, XP000866531 figure 1 | 1-19 |

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